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EGELRUD, Torbjorn

<120> SCCE MODIFIED TRANSGENIC MAMMALS AND THEIR USE AS MODELS OF HUMAN DISEASE

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Gly Pro Leu Val Cys Asn Asp  
 20

<210> 9  
 <211> 38  
 <212> PRT  
 <213> Bos Taurus

<400> 9

Gln Glu Asp Gln Gly Asn Lys Ser Gly Glu Lys Ile Ile Asp Gly Val  
 1 5 10 15

Pro Cys Pro Arg Gly Ser Gln Pro Trp Gln Val Ala Leu Leu Lys Gly  
 20 25 30

Ser Gln Leu His Cys Gly  
 35

<210> 10  
 <211> 37  
 <212> PRT  
 <213> Sus scrofa

<400> 10

Gln Glu Gly Gln Asp Lys Ser Gly Glu Lys Ile Ile Asp Gly Val Pro  
 1 5 10 15

Cys Pro Gly Gly Ser Arg Pro Trp Gln Val Ala Leu Leu Lys Gly Asn  
 20 25 30

Gln Leu His Cys Gly  
 35

<210> 11  
 <211> 34  
 <212> PRT  
 <213> Homo sapiens

<400> 11

Glu Glu Ala Gln Gly Asp Lys Ile Ile Asp Gly Ala Pro Cys Ala Arg  
 1 5 10 15

Gly Ser His Pro Trp Gln Val Ala Leu Leu Ser Gly Asn Gln Leu His  
                   20                  25                  30

Cys Gly

<210> 12  
 <211> 31  
 <212> PRT  
 <213> Rattus norvegicus

<400> 12

Gln Gly Glu Arg Ile Ile Asp Gly Tyr Lys Cys Lys Glu Gly Ser His  
 1                  5                  10                  15

Pro Trp Gln Val Ala Leu Leu Lys Gly Asp Gln Leu His Cys Gly  
                   20                  25                  30

<210> 13  
 <211> 31  
 <212> PRT  
 <213> Mus musculus

<400> 13

Gln Gly Glu Arg Ile Ile Asp Gly Ile Lys Cys Lys Glu Gly Ser His  
 1                  5                  10                  15

Pro Trp Gln Val Ala Leu Leu Lys Gly Asn Gln Leu His Cys Gly  
                   20                  25                  30

<210> 14  
 <211> 7  
 <212> PRT  
 <213> Artificial Sequence

<220>  
 <223> Consensus sequence for cleavage site in C-terminal of SCCE.

<220>  
 <221> misc\_feature  
 <222> (2)..(2)  
 <223> Xaa is either aspartate (Asp) or glutamate (Glu)

<220>  
 <221> misc\_feature  
 <222> (3)..(3)  
 <223> Xaa is either lysine (Lys) or arginine (Arg)

<400> 14

Gly Xaa Xaa Ile Ile Asp Gly  
1 5

<210> 15

<211> 8

<212> PRT

<213> Artificial Sequence

<220>

<223> Consensus of the substrate specificity pouch

<220>

<221> misc\_feature

<222> (1)..(1)

<223> Xaa is any amino acid residue

<220>

<221> misc\_feature

<222> (3)..(3)

<223> Xaa is any amino acid residue

<220>

<221> misc\_feature

<222> (4)..(4)

<223> Xaa is Cys

<220>

<221> misc\_feature

<222> (5)..(5)

<223> Xaa is any amino acid residue

<220>

<221> misc\_feature

<222> (6)..(6)

<223> Xaa is Gly

<220>

<221> misc\_feature

<222> (7)..(7)

<223> Xaa is Asp

<400> 15

Xaa Asn Xaa Xaa Xaa Xaa Xaa Ser  
1 5

<210> 16

<211> 20

<212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM3300

<400> 16  
 ggtggccctg ctcaagtggca 20

<210> 17  
 <211> 22  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM3301

<400> 17  
 caccatggat gacacagcct gg 22

<210> 18  
 <211> 20  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM3302

<400> 18  
 aataaagaaa cacaaaaccc 20

<210> 19  
 <211> 18  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM3418

<400> 19  
 tgtaatatca ttgtgggc 18

<210> 20  
 <211> 18  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM4118

<400> 20  
 ggatgtgaag ctcatctc 18

<210> 21  
 <211> 18  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM4121

<400> 21  
 tggagtcggg gatgccag 18

<210> 22  
 <211> 25  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM4720

<400> 22  
 gggaggggtgg agagagagtg cagtg 25

<210> 23  
 <211> 21  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer SYM4899

<400> 23  
 agtctaggct gcagccccta c 21

<210> 24  
 <211> 22  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer hEXON1

<400> 24  
 ctcgagggat ctgatgtgat cc 22

<210> 25  
 <211> 24  
 <212> DNA  
 <213> Artificial Sequence

<220>  
 <223> PCR primer mEXON1

<400> 25  
 ctgggagtga cttggcgtgg ctct 24

<210> 26  
 <211> 23  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> PCR primer specific for human SCCE IE2  
  
 <400> 26  
 gctctcccat tagtccccag aga 23  
  
 <210> 27  
 <211> 24  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> PCR primer specific for human SCCE MJ2  
  
 <400> 27  
 ccacttggtg aacttgaca cttg 24  
  
 <210> 28  
 <211> 18  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> Forward primer covering the position 427-444 of the human SCCE cDNA sequence.  
  
 <400> 28  
 gcgaaccccc tggaacaa 18  
  
 <210> 29  
 <211> 21  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> Reverse primer covering the position 490-510 of the human cDNA sequence in exon five.  
  
 <400> 29  
 acatccacgc acatgaggtc a 21  
  
 <210> 30  
 <211> 29  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>



<223> The real time amplification probe covering the position 445-473 of the human cDNA sequence in exon four.

<400> 30  
cctgtactgt ctccggctgg ggcactacc 29

<210> 31  
<211> 25  
<212> DNA  
<213> Artificial Sequence

<220>  
<223> PCR primer mS3

<400> 31  
caaggagaaa ggattataga tggct 25

<210> 32  
<211> 21  
<212> DNA  
<213> Artificial Sequence

<220>  
<223> PCR primer 698

<400> 32  
aaggctccgc acccatggca g 21

<210> 33  
<211> 26  
<212> DNA  
<213> Artificial Sequence

<220>  
<223> PCR primer 696

<400> 33  
tgcaatggtg actcaggggg gccctt 26

<210> 34  
<211> 23  
<212> DNA  
<213> Artificial Sequence

<220>  
<223> PCR primer H2

<400> 34  
gacccaggcg tctacactca agt 23

<210> 35  
<211> 25  
<212> DNA

<213> Artificial Sequence

<220>

<223> PCR primer mS4

<400> 35

gagacatga aaacccatcg ctaac

25

<210> 36

<211> 26

<212> DNA

<213> Artificial Sequence

<220>

<223> PCR primer KO 0905

<400> 36

tgactttctt cacactggac gacagc

26

<210> 37

<211> 26

<212> DNA

<213> Artificial Sequence

<220>

<223> PCR primer GR 0905

<400> 37

cttcacactg gctgatagcc tggccg

26

<210> 38

<211> 28

<212> DNA

<213> Artificial Sequence

<220>

<223> PCR primer Ngr

<400> 38

caggggtggcg gaatgacctc atggccct

28

<210> 39

<211> 28

<212> DNA

<213> Artificial Sequence

<220>

<223> PCR primer RA 1016

<400> 39

ctactccaca aggacccatg tcaatgac

28

<210> 40

<211> 28  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> PCR primer nRA 1016  
  
 <400> 40  
 gctgtgtgct ggcattcccg actctaag 28

<210> 41  
 <211> 30  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> SMART II oligonucleotide  
  
 <400> 41  
 aagcagtggg aacaacgcag agtacgcggg 30

<210> 42  
 <211> 27  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> 5'-RACE cDNA synthesis primer  
  
 <220>  
 <221> misc\_feature  
 <222> (27)..(27)  
 <223> n is a or g or c or t  
  
 <400> 42  
 tttttttttt tttttttttt tttttvn 27

<210> 43  
 <211> 45  
 <212> DNA  
 <213> Artificial Sequence  
  
 <220>  
 <223> Long universal primer  
  
 <400> 43  
 ctaatacgac tcactatagg gcaagcagtg gtaacaacgc agagt 45

<210> 44  
 <211> 23  
 <212> DNA  
 <213> Artificial Sequence

&lt;220&gt;

&lt;223&gt; Short universal primer

&lt;400&gt; 44

ctaatacgac tcactatagg gcc

23

&lt;210&gt; 45

&lt;211&gt; 23

&lt;212&gt; DNA

&lt;213&gt; Artificial Sequence

&lt;220&gt;

&lt;223&gt; Nested universal primer

&lt;400&gt; 45

aagcagtgggt aacaacgcag agt

23

&lt;210&gt; 46

&lt;211&gt; 243

&lt;212&gt; PRT

&lt;213&gt; Artificial Sequence

&lt;220&gt;

&lt;223&gt; Deduced amino acid sequence from the C-terminal part of SCCE from cow.

&lt;400&gt; 46

Met	Thr	Thr	Pro	Leu	Val	Ile	Leu	Leu	Leu	Thr	Phe	Ala	Leu	Gly	Ser
1				5				10						15	

Val	Ala	Gln	Glu	Asp	Gln	Gly	Asn	Lys	Ser	Gly	Glu	Lys	Ile	Ile	Asp
		20					25						30		

Gly	Val	Pro	Cys	Pro	Arg	Gly	Ser	Gln	Pro	Trp	Gln	Val	Ala	Leu	Leu
		35					40					45			

Lys	Gly	Ser	Gln	Leu	His	Cys	Gly	Gly	Val	Leu	Leu	Asn	Glu	Gln	Trp
	50					55				60					

Val	Leu	Thr	Ala	Ala	His	Cys	Met	Asn	Glu	Tyr	Asn	Val	His	Met	Gly
65					70					75					80

Ser	Val	Arg	Leu	Val	Gly	Gly	Gln	Lys	Ile	Lys	Ala	Thr	Arg	Ser	Phe
			85					90						95	

Arg	His	Pro	Gly	Tyr	Ser	Thr	Gln	Thr	His	Ala	Asn	Asp	Leu	Met	Leu
			100					105					110		

Val Lys Leu Asn Gly Arg Ala Lys Leu Ser Ser Ser Val Lys Lys Val  
 115 120 125

Asn Leu Pro Ser His Cys Asp Pro Pro Gly Thr Met Cys Thr Val Ser  
 130 135 140

Gly Trp Gly Thr Thr Thr Ser Pro Asp Val Thr Phe Pro Gly Gln Leu  
 145 150 155 160

Met Cys Thr Asp Val Lys Leu Ile Ser Pro Gln Asp Cys Arg Lys Val  
 165 170 175

Tyr Lys Asp Leu Leu Gly Asp Ser Met Leu Cys Ala Gly Ile Pro Asn  
 180 185 190

Ser Arg Thr Asn Ala Cys Asn Gly Asp Ser Gly Gly Pro Leu Met Cys  
 195 200 205

Lys Gly Thr Leu Gln Gly Val Val Ser Trp Gly Ser Phe Pro Cys Gly  
 210 215 220

Gln Pro Asn Asp Pro Gly Val Tyr Thr Gln Val Cys Lys Tyr Val Asn  
 225 230 235 240

Trp Ile Lys

<210> 47

<211> 249

<212> PRT

<213> Artificial Sequence

<220>

<223> Deduced amino acid sequence from the C-terminal part of SCCE from pig.

<400> 47

Met Ala Arg Pro Leu Leu Pro Pro Arg Leu Ile Leu Leu Leu Ser Leu  
 1 5 10 15

Ala Leu Gly Ser Ala Ala Gln Glu Gly Gln Asp Lys Ser Gly Glu Lys  
 20 25 30

Ile Ile Asp Gly Val Pro Cys Pro Gly Gly Ser Arg Pro Trp Gln Val  
 35 40 45

Ala Leu Leu Lys Gly Asn Gln Leu His Cys Gly Gly Val Leu Val Asn  
 50 55 60

Gln Gln Trp Val Leu Thr Ala Ala His Cys Met Met Asn Asp Tyr Asn  
 65 70 75 80

Val His Leu Gly Ser Asp Arg Leu Asp Asp Arg Lys Gly Gln Lys Ile  
 85 90 95

Arg Ala Met Arg Ser Phe Arg His Pro Gly Tyr Ser Thr Gln Thr His  
 100 105 110

Val Asn Asp Leu Met Leu Val Lys Leu Ser Arg Pro Ala Arg Leu Ser  
 115 120 125

Ala Ser Val Lys Lys Val Asn Leu Pro Ser Arg Cys Glu Pro Pro Gly  
 130 135 140

Thr Thr Cys Thr Val Ser Gly Trp Gly Thr Thr Thr Ser Pro Asp Val  
 145 150 155 160

Thr Phe Pro Ala Asp Leu Met Cys Thr Asp Val Lys Leu Ile Ser Ser  
 165 170 175

Gln Asp Cys Lys Lys Val Tyr Lys Asp Leu Leu Gly Ser Ser Met Leu  
 180 185 190

Cys Ala Gly Ile Pro Asn Ser Lys Thr Asn Ala Cys Asn Gly Asp Ser  
 195 200 205

Gly Gly Pro Leu Val Cys Lys Gly Thr Leu Gln Gly Leu Val Ser Trp  
 210 215 220

Gly Thr Phe Pro Cys Gly Gln Pro Asn Asp Pro Gly Val Tyr Thr Gln  
 225 230 235 240

Val Cys Lys Tyr Ile Asp Trp Ile Asn  
 245

<210> 48

<211> 253

<212> PRT

<213> Artificial Sequence

&lt;220&gt;

<223> Deduced amino acid sequence from the C-terminal part of SCCE from  
homo sapiens.

&lt;400&gt; 48

Met Ala Arg Ser Leu Leu Leu Pro Leu Gln Ile Leu Leu Leu Ser Leu  
1 5 10 15

Ala Leu Glu Thr Ala Gly Glu Glu Ala Gln Gly Asp Lys Ile Ile Asp  
20 25 30

Gly Ala Pro Cys Ala Arg Gly Ser His Pro Trp Gln Val Ala Leu Leu  
35 40 45

Ser Gly Asn Gln Leu His Cys Gly Gly Val Leu Val Asn Glu Arg Trp  
50 55 60

Val Leu Thr Ala Ala His Cys Lys Met Asn Glu Tyr Thr Val His Leu  
65 70 75 80

Gly Ser Asp Thr Leu Gly Asp Arg Arg Ala Gln Arg Ile Lys Ala Ser  
85 90 95

Lys Ser Phe Arg His Pro Gly Tyr Ser Thr Gln Thr His Val Asn Asp  
100 105 110

Leu Met Leu Val Lys Leu Asn Ser Gln Ala Arg Leu Ser Ser Met Val  
115 120 125

Lys Lys Val Arg Leu Pro Ser Arg Cys Glu Pro Pro Gly Thr Thr Cys  
130 135 140

Thr Val Ser Gly Trp Gly Thr Thr Thr Ser Pro Asp Val Thr Phe Pro  
145 150 155 160

Ser Asp Leu Met Cys Val Asp Val Lys Leu Ile Ser Pro Gln Asp Cys  
165 170 175

Thr Lys Val Tyr Lys Asp Leu Leu Glu Asn Ser Met Leu Cys Ala Gly  
180 185 190

Ile Pro Asp Ser Lys Lys Asn Ala Cys Asn Gly Asp Ser Gly Gly Pro  
195 200 205

Leu Val Cys Arg Gly Thr Leu Gln Gly Leu Val Ser Trp Gly Thr Phe  
 210 215 220

Pro Cys Gly Gln Pro Asn Asp Pro Gly Val Tyr Thr Gln Val Cys Lys  
 225 230 235 240

Phe Thr Lys Trp Ile Asn Asp Thr Met Lys Lys His Arg  
 245 250

<210> 49

<211> 226

<212> PRT

<213> Artificial Sequence

<220>

<223> Deduced amino acid sequence from the C-terminal part of SCCE from  
 rat.

<400> 49

Met Gly Val Trp Leu Leu Ser Leu Leu Thr Val Leu Leu Ser Leu Ala  
 1 5 10 15

Leu Glu Thr Ala Gly Gln Gly Glu Arg Ile Ile Asp Gly Tyr Lys Cys  
 20 25 30

Lys Glu Gly Ser His Pro Trp Gln Val Ala Leu Leu Lys Gly Asp Gln  
 35 40 45

Leu His Cys Gly Gly Val Leu Val Gly Glu Ser Trp Val Leu Thr Ala  
 50 55 60

Ala His Cys Lys Met Gly Gln Tyr Thr Val His Leu Gly Ser Asp Lys  
 65 70 75 80

Ile Glu Asp Gln Ser Ala Gln Arg Ile Lys Ala Ser Arg Ser Phe Arg  
 85 90 95

His Pro Gly Tyr Ser Thr Arg Thr His Val Asn Asp Ile Met Leu Val  
 100 105 110

Lys Met Asp Lys Pro Val Lys Met Ser Asp Lys Val Gln Lys Val Lys  
 115 120 125

Leu Pro Asp His Cys Glu Pro Pro Gly Thr Leu Cys Thr Val Ser Gly  
 130 135 140



Trp Gly Thr Thr Thr Ser Pro Asp Val Thr Phe Pro Ser Asp Leu Met  
 145 150 155 160

Cys Ser Asp Val Lys Leu Ile Ser Ser Gln Glu Cys Lys Lys Val Tyr  
 165 170 175

Lys Asp Leu Leu Gly Lys Thr Met Leu Cys Ala Gly Ile Pro Asp Ser  
 180 185 190

Lys Thr Asn Thr Cys Asn Gly Asp Ser Gly Gly Pro Leu Val Cys Asn  
 195 200 205

Asp Thr Leu Gln Gly Leu Val Ser Trp Gly Thr Tyr Pro Cys Gly Gln  
 210 215 220

Pro Asn  
 225

<210> 50  
 <211> 249  
 <212> PRT  
 <213> Artificial Sequence

<220>  
 <223> Deduced amino acid sequence from the C-terminal part of SCCE from mouse.

<400> 50

Met Gly Val Trp Leu Leu Ser Leu Ile Thr Val Leu Leu Ser Leu Ala  
 1 5 10 15

Leu Glu Thr Ala Gly Gln Gly Glu Arg Ile Ile Asp Gly Ile Lys Cys  
 20 25 30

Lys Glu Gly Ser His Pro Trp Gln Val Ala Leu Leu Lys Gly Asn Gln  
 35 40 45

Leu His Cys Gly Gly Val Leu Val Asp Lys Tyr Trp Val Leu Thr Ala  
 50 55 60

Ala His Cys Lys Met Gly Gln Tyr Gln Val Gln Leu Gly Ser Asp Lys  
 65 70 75 80

Ile Gly Asp Gln Ser Ala Gln Lys Ile Lys Ala Thr Lys Ser Phe Arg  
 85 90 95

His Pro Gly Tyr Ser Thr Lys Thr His Val Asn Asp Ile Met Leu Val  
 100 105 110

Arg Leu Asp Glu Pro Val Lys Met Ser Ser Lys Val Glu Ala Val Gln  
 115 120 125

Leu Pro Glu His Cys Glu Pro Pro Gly Thr Ser Cys Thr Val Ser Gly  
 130 135 140

Trp Gly Thr Thr Thr Ser Pro Asp Val Thr Phe Pro Ser Asp Leu Met  
 145 150 155 160

Cys Ser Asp Val Lys Leu Ile Ser Ser Arg Glu Cys Lys Lys Val Tyr  
 165 170 175

Lys Asp Leu Leu Gly Lys Thr Met Leu Cys Ala Gly Ile Pro Asp Ser  
 180 185 190

Lys Thr Asn Thr Cys Asn Gly Asp Ser Gly Gly Pro Leu Val Cys Asn  
 195 200 205

Asp Thr Leu Gln Gly Leu Ala Ser Arg Gly Thr Tyr Pro Cys Gly Gln  
 210 215 220

Pro Asn Asp Pro Gly Val Tyr Thr Gln Val Cys Lys Tyr Lys Arg Trp  
 225 230 235 240

Val Met Glu Thr Met Lys Thr His Arg  
 245